



## PRODH gene

proline dehydrogenase 1

### Normal Function

The *PRODH* gene provides instructions for producing the enzyme proline oxidase (also known as proline dehydrogenase), which is found primarily in the brain, liver, and kidney. Within cells of these organs, this enzyme functions in energy-producing structures called mitochondria.

Proline oxidase begins the process of breaking down the protein building block (amino acid) proline by starting the reaction that converts it to pyrroline-5-carboxylate. A subsequent step converts this intermediate product to the amino acid glutamate. The conversion between proline and glutamate is important in maintaining a supply of the amino acids needed for protein production, and for energy transfer within the cell.

### Health Conditions Related to Genetic Changes

#### hyperprolinemia

At least 15 mutations in the *PRODH* gene have been found to reduce the activity of the proline oxidase enzyme. These mutations substitute one amino acid for another amino acid in the enzyme, causing it to perform its function in proline breakdown (degradation) less efficiently. A reduction in proline oxidase function results in a buildup of proline in the body, and in severe cases of hyperprolinemia can cause seizures, intellectual disability, or other neurological or psychiatric problems.

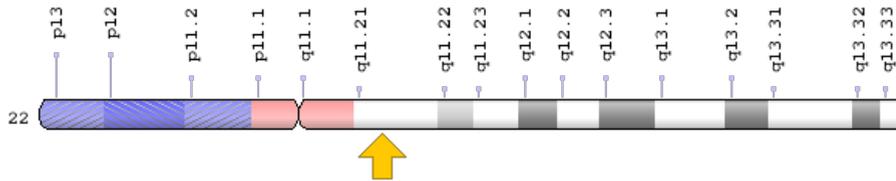
#### other disorders

Several studies have shown an association between variations in the *PRODH* gene and psychiatric disorders such as schizophrenia, while others have shown no significant association. Most of the variations in the *PRODH* gene result in the substitution of one amino acid for another in the proline dehydrogenase enzyme. The amino acid substitution reduces the enzyme's activity, resulting in less efficient breakdown of proline. Researchers believe that elevated proline levels may affect the action of certain chemicals that transmit signals between neurons in the brain (neurotransmitters), resulting in an increased risk of psychiatric disorders.

## Chromosomal Location

Cytogenetic Location: 22q11.21, which is the long (q) arm of chromosome 22 at position 11.21

Molecular Location: base pairs 18,912,774 to 18,936,553 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- FLJ33744
- HSPOX2
- MGC148078
- MGC148079
- p53 induced protein
- PIG6
- PROD\_HUMAN
- PRODH1
- PRODH2
- proline dehydrogenase (oxidase) 1
- proline dehydrogenase (proline oxidase )
- proline oxidase 2
- Proline oxidase, mitochondrial
- SCZD4
- TP53I6
- tumor protein p53 inducible protein 6

## Additional Information & Resources

### Educational Resources

- Sequence-Evolution-Function (2003): Proline Biosynthesis  
<https://www.ncbi.nlm.nih.gov/books/NBK20266/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28PRODH%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

### OMIM

- PROLINE DEHYDROGENASE (OXIDASE) 1  
<http://omim.org/entry/606810>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_PRODH.html](http://atlasgeneticsoncology.org/Genes/GC_PRODH.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=PRODH%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=9453](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9453)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/5625>
- UniProt  
<http://www.uniprot.org/uniprot/O43272>

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